

Suggested Follow-up for Galactosemia Deficient GALT with Normal Total Galactose

Possible Causes:

Classic galactosemia is a condition of abnormal galactose metabolism caused by deficient functioning of galactose-1-P-uridyl transferase (GALT). When GALT is moderately deficient, the infant may have **Duarte galactosemia** or maybe a **carrier** for a form of galactosemia.

GALT can also be falsely deficient if specimen is exposed to extremes of heat and/or humidity.

Next Steps if Abnormal:

See infant as soon as possible to ascertain health status. Consult a pediatric metabolic specialist and initiate diagnostic evaluation and treatment as recommended. If the mother is breast-feeding, it is not necessary to switch to soy-based infant formula since the total galactose is within normal limits.

Repeat GALT and total galactose screening on filter paper and send to the DHEC laboratory. If GALT is still deficient in repeat specimen, further testing is necessary to clarify the diagnosis.

Neonatal Presentation:

Classic galactosemia - hypoglycemia, jaundice, sepsis, failure to thrive.
Duarte galactosemia - none.

Emergency Treatment: Usually none.

Standard Treatment:

Classic galactosemia - galactose restricted diet for life.
Duarte galactosemia - usually none past infancy.

Advice for Family:

Provide basic information about classical galactosemia. The handout, *When Baby Needs a Second Test for Classic Galactosemia*, may be used for this purpose. Stress the importance of seeking immediate medical attention if the infant shows any signs of illness.

Internet resources:

<http://ghr.nlm.nih.gov/condition/galactosemia>

https://www.newbornscreening.info/Parents/otherdisorders/Duarte_galactosemia.html